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Foundation for Ichthyosis & Related Skin Types

Ichthyosis Focus

A Journal for Friends of FIRST

SPRING 2025 • Volume 44, No. 1

MINDFULNESS MATTERS

Techniques for improved mental health....6

Strong Foundation, Exciting Future

It's an honor to step into the role of Chairman of the Board at FIRST. I'm thrilled to partner with our dedicated Board of Directors, Medical and Scientific Advisory Board (MSAB), volunteers and staff to continue serving this incredible community.

Sean McTernan

Our journey with FIRST began nine years ago, when our youngest daughter, Claire, was born with a collodion membrane and later diagnosed with ARCI temperature-sensitive lamellar ichthyosis. In those early, uncertain days, FIRST was one of the few places we could turn for answers, perspective, and hope.

That early support made a lasting impact—carving a special place in my and my family's hearts and deepening our gratitude for FIRST and this community. It also gave us a firsthand appreciation for the dedication of FIRST's volunteers and staff. This support is what led me to join the board seven years ago.

Since then, I've had the privilege of working alongside outstanding board chairs—Jeff Hoerle, Mark Evans, Tracie Pretak, and most recently, Beth Hampshire—whose leadership has helped make FIRST the strong, mission-driven organization it is today.

Looking ahead, I'm especially excited about what's to come:

- **A new strategic plan**—coming later this year—focused on patient empowerment as well as access to experts, dermatology training, referral networks, and treatment innovation.
- **A new Research Roadmap**, which outlines how FIRST and the MSAB will support scientific progress through partnerships, clinical trials development and participation, collaboration with academic research and for-profit biopharma, and drug approval advocacy. As MSAB Chair, Dr. Amy Paller put it: "This will be the decade of discovery" — (read more on page 9) and FIRST is ready to play a meaningful role during this exciting time.
- **Growing member engagement**, through local support forums and our June 2026 National Conference in Minneapolis. I hope to see many of you there.

To our board, MSAB, staff, and volunteers—thank you. I look forward to keeping you updated and working together to continue to push this mission forward.

Sincerely,

Sean McTernan

Vol. 44, No. 1 Spring 2025

FIRST exists to improve lives and seek cures for those affected with ichthyosis or a related skin type.

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One-Day Forums Bring Support Close to Home

Patient Support Forums are regional events that bring together affected families and medical experts for a day of education and connection. The following events are planned for 2025:

Austin, TX - May 17
CT - TBD
Stanford, CA - TBD
Detroit, MI - TBD

Learn more and register at firstskinfoundation.org/patient-support-forums.

New Faces Join Board of Directors

Evan Westlake

Sherman, Illinois
Parent of affected daughter

I'm a proud husband to Courtney and father to Connor and Brenna. We live in central Illinois and love to travel and spend time outdoors. Professionally, I work in finance for a regional construction company, following many years in banking.

My connection to FIRST began in 2011 when Brenna was born with harlequin ichthyosis. Like so many others, we were suddenly thrust into the ichthyosis community and had to learn everything we could, as quickly as we could. FIRST was an invaluable resource for us and our medical team in those early days, and I will always be grateful for that support.

Since 2014, we've attended every National Conference and have loved watching this incredible community grow and thrive. I'm excited to serve on the board to help strengthen connections and give back to a community that has meant so much to our family.

Also joining the board is Kara McCafferty of Los Angeles.

Dr. Bunick Named Top Editor of Dermatology Times

Congratulations to Christopher Bunick, MD, PhD, who was recently named editor in chief of Dermatology Times.

Bunick serves as a member of FIRST's Medical Scientific Advisory Board and The Research Committe. Learn more about his career and interests at dermatologytimes.com/view/introducing-dermatology-times-2025-editor-in-chief.

Dermatology TIMES

Heart Rate Monitors: A Tool for Preventing Overheating

Heart rate monitors have been around for a while. But have you ever considered them as a tool to help manage and prevent overheating?

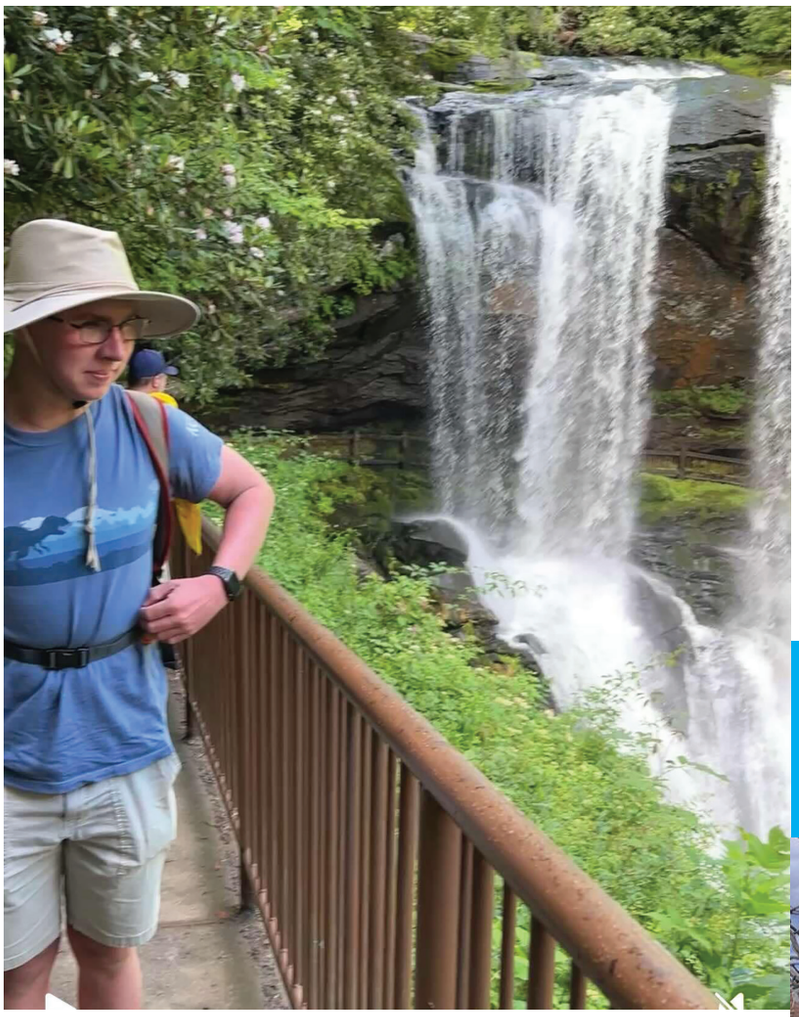
Put simply: your heart rate will tell you how close you are to overheating.

Traditionally, the risk of overheating has been difficult to anticipate and prevent, leading to anxiety, reduced activity, and feeling a bit out of control. At worst, the risk of overheating can produce emergency situations.

"In 2023, I joined the FIRST patients and family members climbing the Grand Canyon – a strenuous hike in a hot, dry, sunny, and unforgiving environment," explains Dr. Cheryl Bayart, pediatric dermatologist and medical advisor for FIRST. "I was so relieved to see many hikers monitoring their heart rates and using the readings to guide pace and hydration. I am very impressed by this intelligent use of technology and how in touch everyone was with their bodies."

What makes heart rate monitoring so promising is that it provides objective data about internal physiological responses that aren't apparent from the skin's surface. Heart rate monitoring provides a window into how hard the cardiovascular system is working – to fuel the body for activity and to keep it cool. Monitors can alert you when your heart rate indicates you may be approaching your heat tolerance threshold, allowing for preventive action before symptoms become severe.

Monitors can alert you when your heart rate indicates you may be approaching your heat tolerance threshold, allowing for preventive action before symptoms become severe.



Ethan Edwards uses a heart rate monitor to track signs of overheating

Using Maximum Heart Rate as a Safety Gauge

Your personal maximum heart rate is often estimated as 220 beats per minute minus your age. This heart rate maximum serves as an important reference point.

Heart rate zones represent different intensity levels based on percentages of your maximum heart rate, with lower zones (around 50 to 60 percent) indicating light activity and higher zones (80 to 90 percent and above) representing more cardiovascular effort. For people with ichthyosis, monitoring which zone coincides with overheating symptoms helps establish personal thresholds for when to implement cooling strategies. Reaching moderate or high zones during minimal activity can be a signal that the body is working harder to regulate temperature.

"What's particularly valuable about using maximum heart rate as a reference is the personalization," notes Dr. Bayart. "Some patients discover they need to take cooling measures when reaching just 65 percent of their maximum, while others might not experience symptoms until 80 percent. These individualized thresholds provide precise guidance that generic advice simply cannot offer."

Getting Started with Heart Rate Monitors

1. Choose a device with continuous heart rate monitoring. Focus on heart rate functionality rather than skin temperature sensing, which may be less reliable for people with ichthyosis.
2. Calculate your estimated maximum heart rate (220bpm minus your age) and note what percentages correspond to different heart rate zones.
3. Establish your baseline readings by wearing the device during regular activities for at least one week. Pay particular attention to what percentage of your maximum heart rate correlates with early feelings of overheating.
4. Explore custom notifications for heart rate spikes.
5. Keep a simple journal recording any instances of feeling overheated alongside your heart rate data to identify personal patterns.
6. Discuss this data with your doctor if you need help developing personalized guidelines for activity modifications.
7. Discuss this data with your family and support team to establish communication protocols for activity and heat exposure.

The more you track, the more you will learn about yourself and the more empowered your choices can be. For example, your heart rate might stay elevated for longer periods of time in humid places versus dry.

While this technology shows promise, experts emphasize these tools should complement, not replace, your existing care regimen. Proper skincare, hydration, appropriate clothing, and environmental modifications remain essential components of ichthyosis care.

SUPPORT A GRAND EXPEDITION

May 2025

FIRST members are tackling the Grand Canyon again this year! Is this strenuous, inspiring trek calling your name? Support a team—affected individuals, family members, or medical staff! The teams fundraise in a friendly competition and keep each other motivated all the way through.

If you'd like to support the hikers, please use the QR code above or this link: givebutter.com/iam25

Practice Mindfulness for Mental Health and Wellbeing

Mikela Murphy

Hi everyone! If you don't know me already, my name is Mikela, and I have ARCI ichthyosis. I'm halfway through my third year in a clinical psychology doctoral program at Fordham University. I wanted to share some information about mindfulness because it's a technique that I use regularly to help when I'm feeling stressed or anxious.

What Is Mindfulness?

Mindfulness is defined as "the awareness that emerges through paying attention on purpose, in the present moment, and nonjudgmentally to the unfolding of experience moment by moment."[1] There are hundreds of mindfulness techniques in existence, but several common techniques that you may be familiar with include breathing exercises and mindfulness meditations. The goal of mindfulness practice is to increase one's contact with the present moment and bring about a sense of relaxation.

Mindfulness can be beneficial for adults, children, and adolescents. Mindfulness can be a useful tool for coping with stress and can help youth and adults dealing with medical illness and treatment.[2–4] Specific mental health conditions have also seen benefits from mindfulness practice in youth and adults including anxiety and depression.[5] Finally, mindfulness practice can also be beneficial for parents and caregivers. Specifically, mindfulness has been shown to improve interactions between parents and children and decrease parents' stress.[6,7]

How to Practice Mindfulness Techniques

Mindfulness practices can range in length from just a few seconds, like taking a few deep breaths, to a full practice, like listening to a mindfulness recording. Start by exploring some of the mindfulness resources on page 15. Practice the technique a few times a week or even daily (parents/caregivers: practice it with your child) so that the skill becomes a habit. It can be helpful to incorporate mindfulness practice into an existing routine or set reminders to practice.

To summarize, mindfulness practices have numerous benefits for adults, adolescents, children, and their parents. They are useful for general wellbeing as well as improving specific mental health concerns. Mindfulness practices can be integrated into your weekly routine, even if you're only practicing for a few seconds at a time. There are numerous mindfulness videos and audio recordings online, I've shared a few on on the next page to get started. Let me know what else would be helpful by emailing info@firstskinfoundation.org.

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Foundation for Ichthyosis & Related Skin Types

FY 2023 FIRST ANNUAL REPORT

About Us

OUR MISSION STATEMENT

Our mission is to improve lives and seek cures for those affected by ichthyosis and related skin types.

OUR LEADERSHIP

The Foundation for Ichthyosis and Related Skin Types (FIRST) is governed by a volunteer Board of Directors dedicated to supporting our mission with their time and talents. FIRST's board members are active participants in the various activities of the Foundation and are representative of our diverse population.

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How We Invest Your Money

We believe our donors deserve full transparency on how their money is allocated in pursuit of our mission "to Improve lives and seek cures for those affected by ichthyosis and related skin types."

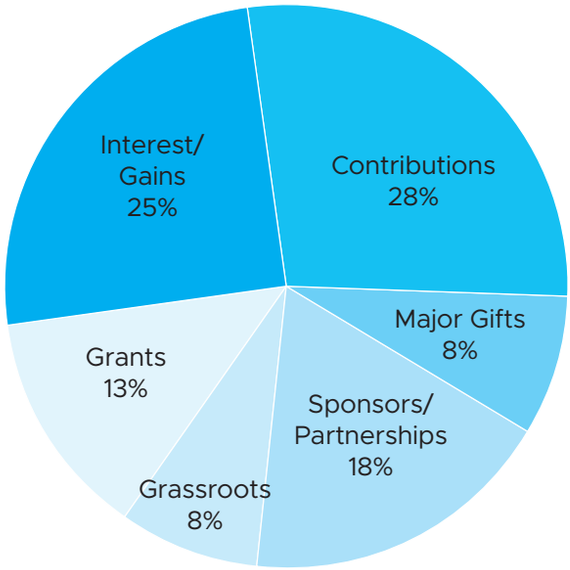
The following pages contain information from our audited financial statements to show how we raised and stewarded donor dollars in fiscal year 2022-23 to empower affected individuals and their families.

REVENUE

Total revenue, gains and other support for the year ended September 30, 2023

	FY '22-'23
Contributions	$248,148
Major Gifts	$66,443
Sponsors/Partnerships	$163,102
Grassroots	$72,213
Grants	$117,500
Other	$217,013
Total Support	**$884,419**

TOTAL SUPPORT



2023 Reach and Results

FIRST hosted member meet-ups in St. Louis, Tampa and Seattle that were attended by **62** individuals and families.

177,918 unique visitors found their way to firstskinfoundation.org for help and information.

310 new families reached out to FIRST for information, resources and to become a part of this nurturing community.

281 people inquired about ichthyosis experts through our Physician Referral program.

TWO researchers were awarded a total of **$100,000** to further their research.

- Dr. Benjamin Nanes from the University of Texas Southwestern Medical Center looked at keratin isoform switching during epidermal differentiation.
- Dr. Cory Simpson's grant studies pathogenic mechanisms and evaluates new treatment approaches for Darier disease.

Dr. Benjamin Nanes

Dr. Cory Simpson

25 members of our community received over **$15,000** in financial support for the purchase of creams and lotions and UFIRST scholarships for college.

FIRST worked with **six** pharmaceutical companies to create patient-focused clinical trials and marketed those trials to our membership to accelerate the development of new therapies.

Member Stories - Hope for Patients

Genetic testing through the Ichthyosis Registry revealed Camillia's rare ichthyosis-associated heart complication, allowing her to receive prompt treatment. Read Camillia's story at firstskinfoundation.org/camellia-2024.

Thanks to a dedicated team at Northwestern Hospital in Chicago focused on researching Darier's disease, Genevieve believes effective treatment – and an ultimate cure – is closer than ever. Read Genevieve's story at firstskinfoundation.org/genevieve-2025.

MSAB Drives Progress on Many Fronts

The Medical and Scientific Advisory Board is swinging into action to move forward the many advances related to ichthyosis. Long an advisory group to FIRST, the MSAB has approximately 25 members, all physician experts on ichthyosis. The goals of the MSAB are to guide FIRST in discovering content for education, partner in helping patients with concerns, promote research that leads to better understanding of ichthyosis and new therapies, and provide information about international and national activities in the field. The MSAB includes some younger physicians who are interested in ichthyosis and thus serves as a way to mentor future leaders in the field.

At the every-other-year FIRST National Conference, MSAB members have been central in giving talks, networking with affected individuals and families, conducting research, and providing one-on-one guidance about diagnoses. They are also a network for physician referral to make the diagnosis of ichthyosis subtypes and provide cutting-edge recommendations. MSAB members have been central to growing the Ichthyosis Registry.

The MSAB meets at least quarterly and presents to the Board of Directors of FIRST on a regular basis. At MSAB meetings, members share information about activities. For example, at the most recent meeting, there was discussion about new guidelines for treatment of ichthyosis that are being published in 2025, a new consortium of sites for research that has been formed and is hoping for funding from

the National Institutes of Health to forge new paths towards treatment, and a proposed re-classification of ichthyosis and related skin types.

The latter has grouped ichthyosis subtypes and all of these "related skin types" in what is now called "epidermal differentiation disorders", simplified to EDDs. For translation, "epidermal" refers to the outer layer of skin where all the action is in ichthyosis and "differentiation" refers to the healthy maturation of skin to form the skin barrier, which is impaired in ichthyosis.

The MSAB is also supporting the development of guidelines through the Pediatric Dermatology Research Alliance for neonatologists, dermatologists, and other specialists to educate and standardize best care for newborns with ichthyosis.

Other actions to look forward to in the next year or two from the MSAB are:

- Creating an inventory of MSAB-approved letters that affected individuals can use (for example, for taking emollients onboard when traveling)
- Annotating the list of dermatologist members to help identify areas of expertise and developing a roster of national subspecialists (beyond dermatologists) who have expertise
- Advising about the new FIRST "Research Roadmap"
- Generating data-based informational short videos for posting on social media about ichthyosis
- Continuing the EDVYCE tool online to answer questions about ichthyosis
- Sharing updates through the website, newsletter, and social media about new guidelines, classification, and therapies
- Being present in full-force at the June 2026 conference in Minneapolis to meet with participants and drive forward education and research

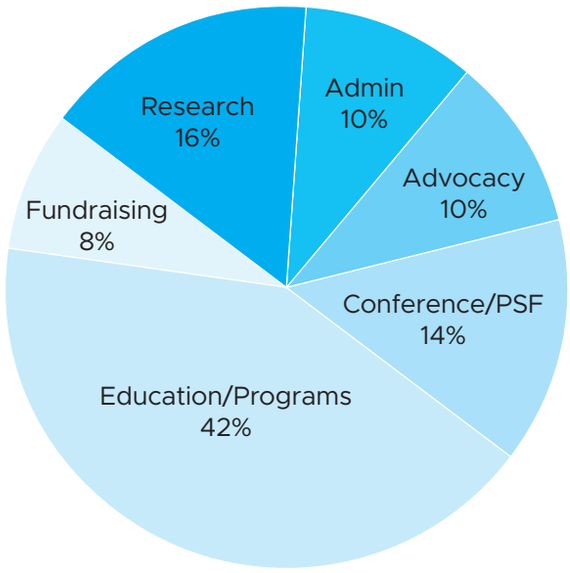
The Impact of Your Donations

EXPENSE ALLOCATION

Total mission program and support expenses for the year ended September 30, 2023, were as follows:

	FY '22-'23
Advocacy	$76,713
Conference/Meet-Up	$105,003
Education/Programs	$321,556
Research	$117,517
Total Mission Program Services	**$620,789**
Management and General	$73,153
Fundraising	$62,147
Total Support Services	**$135,300**
Total Mission and Support Services	**$756,089**

YOUR IMPACT



Sustainability for Our Future

FIRST prudently invests reserves in a well-diversified portfolio to ensure consistent delivery of services regardless of the economy or investment environment. It is our goal to maintain one year of operating expenses in reserves.

NET ASSETS

	2023				
	Without Donor Restrictions	Temporarily Restricted	Permanently Restricted	2023 Totals	2022 Totals
Support and operating revenue					
Contributions	$566,871	$32,222	$77,338	$676,431	$619,447
Net assets released from restrictions	$110,171	-$110,171	$0	$0	$0
Total support and revenue	**$677,042**	**-$77,949**	**$77,338**	**$676,431**	**$619,447**
Operating expenses					
Program services	$620,790	$0	$0	$620,790	$695,120
Management and general support	$73,153	$0	$0	$73,153	$112,637
Fundraising support	$62,147	$0	$0	$62,147	$83,924
Total operating expenses	**$756,091**	**$0**	**$0**	**$756,091**	**$891,681**
Changes in net assets before non-operating revenues and expenses	-$79,049	-$77,949	$77,338	-$79,660	-$272,234
Non-operating revenues and expenses					
Investment income (loss)	$207,988	$0	$0	$207,988	-$387,094
Change in net assets	**$128,939**	**-$77,949**	**$77,338**	**$128,328**	**-$659,328**
Net assets, beginning of year	**$1,043,955**	**$721,864**	**$160,512**	**$1,926,331**	**$2,585,659**
Net assets, end of year	**$1,172,894**	**$643,915**	**$237,850**	**$2,054,659**	**$1,926,331**

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Practice Mindfulness for Mental Health and Wellbeing

Breathing technique (adult version)

Breathing technique (child version)

5 senses grounding exercise (adult version)

5 senses grounding exercise (child version)

Yoga Teachers With Ichthyosis Offer Classes Online

FIRST volunteers Sacha Schenker and Mui Thomas offer live classes and recordings, for free and open to all ages and skill levels. Classes focus on mindfulness and include movement, meditation, and breathwork...starting at just 15 minutes! Scan the QR code to register for upcoming classes or view recordings.

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AAD 'Think Like an Expert' Session Informs Wider Dermatology Community

A session titled "Think Like an Expert About Ichthyosis" was held recently at the 2025 American Academy of Dermatology Annual Meeting in Orlando, FL. In its second year, this session serves to bring attention to this rare condition and increase knowledge among dermatologists encountering ichthyosis sporadically in their practice.

Three physicians with research interests in ichthyosis shared updates related to the ways we think about ichthyosis and how to treat it.

Dr. Amy Paller, chair of FIRST's Medical and Scientific Advisory Board, presented a new naming classification that is being introduced to the scientific and practicing community. The family of conditions will fall under the umbrella called Epidermal Differentiation Disorders (EDD). The new system will include disorders not previously included within the ichthyoses such as Darier's disease. The new system includes the gene mutation and seeks to eliminate derogatory names like "ichthyosis", "hystrix", and "vulgaris" while also doing away with eponyms like "Netherton" and "Gaucher".

The new classification recognizes that the best new therapies being developed are linked to knowledge

Dr. Keith Choate

of the origin and development of the condition. The new names promote the need for increased molecular analysis, which will improve the chance for successful therapies.

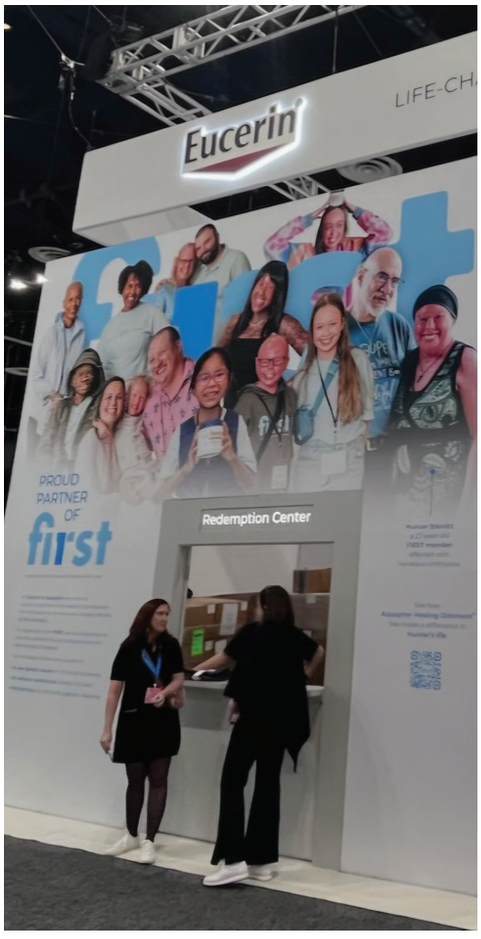
Dr. Keith Choate presented an update on the Yale Ichthyosis Registry. Genetic diagnosis have been provided to 1,449 people and samples are pre-screened for mutations in 72 genes. At this time, 15 percent of the samples received are still "in discovery" or not as easily identified. Dr. Choate shared he is working on a trial for a promising new treatment for Hailey-Hailey.

Juliette Mazereeuw-Hautier presented results of a new study measuring the effectiveness of biologics within EDD. Not unusually, the study pointed to the need for further study. The results were mixed with biologics having some positive effect in approximately half of those studied. Frustrating to researchers and clinicians, the effects were unpredictable. Sometimes the effect was moderate or did not last long. In others, some symptoms worsened. An additional drawback is the high cost of the treatment.

The session also included two patient advocates sharing their life experiences with dermatologists who may not encounter patients with ichthyosis in their practice.

- FIRST Development Consultant Denise Gass spoke of living in a family with three other affected individuals and growing up with the time-consuming burden of near constant skin care. She also offered non-clinical advice to doctors on how the best care for their patients with these conditions.
- Janice Schwartz, executive director of PC Project, talked about her life living with Pachyonychia Congenita, and a new treatment she has found to be very helpful in managing her symptoms.

FIRST continues to seek ways to increase its presence and raise awareness of the unique needs of our ichthyosis community among this important audience.



Dr. Paller Declares 'Decade of Discovery and Drug Development' for Ichthyosis

Dr. Paller

The ichthyosis community gathered on Jan. 25 for Northwestern University's Winter Warm Up, drawing people from Canada to Indiana and surrounding areas.

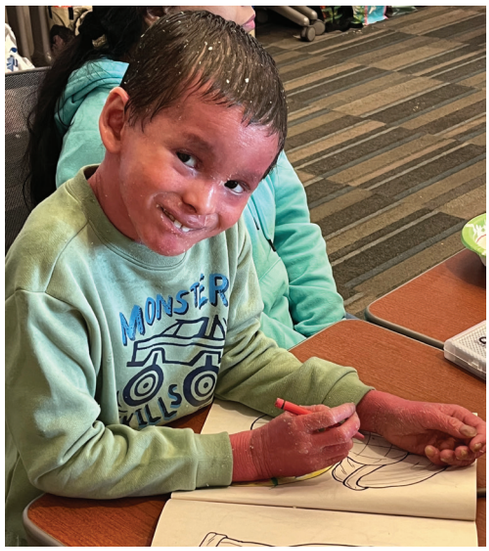
The afternoon event was held at Lurie Children's Hospital in downtown Chicago and was the 10th annual meeting hosted by Dr. Amy Paller.

Dr. Paller gave an update on pharmaceutical research, advocacy, and academic research. Dr. Paller pointed to a strong foundation of genetic diagnoses for more than 1,400 people in the National Registry for Ichthyosis and Related Skin Types. Added to that, she noted interest from several pharmaceutical companies currently in various stages of drug development. For these reasons she declared 2025 to 2035 the "Decade of Discovery and Drug Development," which brought cheers from the crowd.

Dr. Paller mentioned an effort to rename ichthyosis and some of its subtypes such as Netherton, vulgaris, and harlequin. She also explained an effort underway from FIRST, sponsored by Beiersdorf, to publish an official document that outlines best practices for care of a newborn with ichthyosis.

The event also included small group discussions in which parents, kids, and affected adults shared experiences and offered advice to one another. The conversation groups also contributed insights for researchers who were present.

Researchers onsite were also collecting tape strip samples to include in their study. Most participants find this to be a simple and painless way to help push research forward. The process involves a clear, circular



sticker applied to the arm, removed, and placed on a research card.

David Artz, a podcaster and teen with IFAP, was recording for his StoryAidHelp Project.

Many thanks to the event's volunteer organizers and sample sponsors for their support.

Including Ichthyosis from First Date to 'I Do'

Submitted by Abby Evans

Like many in their 20s, I struggled with dating, especially since I graduated college in 2020 during a global shutdown. Instead, I focused on my career, and my friends, and my family. I lived with my best friend from college, so I never felt overly lonely or had too much time to long for a relationship. I stayed occupied with my busy social life, graduate school, and my family.

However, somewhere along the way, I created a dating app profile. Even though I was surrounded by so many friends and family, I knew I wanted to find a partner to share life with. I must admit I didn't think much of my ichthyosis when I created the profile. I knew it would have to come up eventually with potential partners, but I was not overly eager to broach the topic.

When I first matched with PJ, we texted for a little while before deciding to meet in person. I was hesitant to mention anything about my ichthyosis, but I started to get very nervous about what could happen if I didn't. My worries included "What kind of lotion should I use so if we hug, I don't get his shirt greasy?" and "What if he wants to sit outside at the restaurant and I overheat?"

Before our first date, PJ asked me what my dealbreakers were in a relationship. I wasn't planning to say anything serious, but he opened up about what he wanted in the future, which made me comfortable being honest with him about my skin condition and needs. Relieving this worry made all the other normal first date worries that much easier to deal with.

After dating for a couple of years, PJ and I got engaged in October 2024 and plan to get married in December 2025. PJ has a keen understanding of what life with ichthyosis is like for me. He knows to get me an ice pack when I have overheated or to grab a tube of lotion for the road when we leave the house. I guess I got lucky and fell into a relationship with the right person for me. PJ has always shown a curiosity and a desire to understand me. He admitted that he tried to research

ichthyosis before our first date so that he would better understand without having to ask me anything. (He said he never found much…. Obviously, he didn't come across the FIRST website!) I never asked him to do any of this, but this care and compassion for me and his work to understand every facet of my life means so much to me.

Throughout our relationship, I have had to advocate and teach him more about me along the way. For instance, I think a big surprise for him was just how much I need to vacuum! About a year into dating, we went to the Caribbean with his family for a vacation and I had to do my fair share of teaching and advocating in terms of tolerating the heat and not overheating. I must advocate for myself, but PJ has also always approached any facet of my life with empathy and compassion -- and that is one of the many reasons why he is the perfect match for me.

I am certain that not everyone would be this compassionate towards me or as forward-thinking to make me comfortable on our first date and throughout our relationship. However, in a way, having ichthyosis and the needs that present with it was a sure way to filter out any partners who were not the right match for me. Ichthyosis certainly made things a little bit more complicated with dating, but I never let it be at the forefront of my mind and once I found the right person, I brought it up and they were able to show me if we were meant to be together.

Research Furthers Understanding of ARCI, SLS

Tofacitinib ameliorates skin inflammation in a patient with severe autosomal recessive congenital ichthyosis

Kennedy Gallagher

Predoctoral fellow at Northwestern University

Reference: Yu-Chen Lin, Yi-Kai Hong, Wilson Jr F Aala, Kiyotaka Hitomi, Masashi Akiyama, John A McGrath, Chao-Kai Hsu

Clinical and Experimental Dermatology, Volume 49, Issue 8, August 2024, Pages 887–892, https://doi.org/10.1093/ced/llae080

Review: Autosomal recessive congenital ichthyosis (ARCI) is a group of scaling disorders that causes dry, scaly skin and abnormal water loss through the skin barrier. Current treatments for this condition are limited and largely not effective. This report highlights the case of a 27-year-old man with ARCI, caused by change in a gene called TGM1, which produces a protein called transglutaminase 1. Transglutaminase 1 is critical in making the skin barrier.

Many people with ARCI have been treated with targeted biologic medications, such as dupilumab, secukinumab, and ustekinumab, that treat eczema or psoriasis. These agents are very specific in their targeting of inflammation – and they have not worked for those with ARCI due to gene changes in TGM1 (often called lamellar ichthyosis). The inflammatory pathways that these medications target require activating Janus kinase (JAK), suggesting that inhibiting JAKs might offer a new treatment approach.

The patient was prescribed a JAK inhibitor called tofacitinib (11 mg daily) for 26 weeks. Within the first month, his skin showed significant improvements, including reduced skin redness, cracking, and itching. His overall quality of life improved. His skin water loss rate decreased at 10 weeks but increased again in the weeks that followed.

This study suggests that oral tofacitinib could be a promising treatment option for patients with ARCI, but it should be noted that: i) this is a single case report and failures of use of a JAK inhibitor are not typically reported; ii) this is a costly medication that is not approved for ARCI; and iii) this group of medications – and particularly tofacitinib – has a boxed warning from the Food and Drug Administration because of severe possible side effects that include malignancy, infections, clots, and cardiovascular disease. Nevertheless, newer JAK inhibitors are available that have not shown these issues to date, suggesting the possibility that they could be trialed in the future.

Accumulation of ether phospholipids in induced pluripotent stem cells and oligodendrocyte-lineage cells established from patients with Sjogren-Larsson syndrome.

Katie Hummel

Predoctoral fellow at Northwestern University

Reference: Yamaguchi Y; Okuno H; Tokuoka S; Kita Y; Sanosaka T; Kohyama J; Kurosawa K; Sakai N; Miya F; Takahashi T; Kosaki K; Okano
Congenital Anomalies. 65(1):e12587, 2025 Jan-Dec.

Review: Sjogren-Larsson Syndrome (SLS) is a rare genetic condition that leads to dry, scaly skin and neurologic issues. It is caused by changes in a gene called ALDH3A2, which is responsible for producing an enzyme called fatty aldehyde dehydrogenase (FALDH). This enzyme helps to break down certain fats in the body. When this enzyme does not work properly, these fats can build up, causing these health problems.

Not much is known about how fat metabolism works in SLS, but previous research on a deceased 65-year-old patient showed an unusual buildup of certain fats in the brain. To learn more, researchers created a disease model using nerve cells taken from two patients with SLS. From these cells, the scientists made "stem cells" by treating the cells with chemicals that made them able to become any cell type in the body.

The study found that these stem cells from both of the patients with SLS had almost no activity for the involved enzyme, FALDH. When the scientists looked at the fats in brain-like cells derived from the stem cells, they found the same type of fat buildup seen in the brain of the deceased patient. This suggests that the stem cell model can be used to better understand how SLS affects the brain and could be helpful for future research examining if a new treatment could help with the neurologic issues.

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